

SEQUENCE LISTING

and B

<110> Daniel H. Cohn Muhammad Faiyaz ul Haque Lily M. King Deborah Krakow

<120> GENETIC MARKER FOR SPONDYLOEPIMETAPHYSEAL DYSPLASIA

<130> 18810-81553

<140> US 09/898,200

<141> 2001-07-02

<150> 09/399,212

<151> 1999-09-17

<160> 33

<170> FastSEQ for Windows Version 4.0

<210> 1

<211> 2014

<212> DNA

<213> Homo sapiens

<400> 1

ctgetgeege egeegeegee geegteeetg egteettegg tetetgetee egggaeeegg 60 ctccgccgca gccagccagc atgtcgggga tcaagaagca aaagacggag aaccagcaga 120 aatccaccaa tgtagtctat caggcccacc atgtgagcag gaataagaga gggcaagtgg 180 ttggaacaag gggtgggttc cgaggatgta ccgtgtggct aacaggtctc tctggtgctg 240 gaaaaacaac gataagtttt gccctggagg agtaccttgt ctcccatgcc atcccttgtt 300 actccctgga tggggacaat gtccgtcatg gccttaacag aaatctcgga ttctctctctg 360 gggacagaga ggaaaatatc cgccggattg ctgaggtggc taagctgttt gctgatgctg 420 gtctggtctg cattaccagc tttatttctc cattcgcaaa ggatcgtgag aatgcccgca 480 aaatacatga atcagcaggg ctgccattct ttgaaatatt tgtagatgca cctctaaata 540 tttgtgaaag cagagacgta aaaggcctct ataaaagggc cagagctggg gagattaaag 600 gatttacagg tattgattct gattatgaga aacctgaaac tcctgagcgt gtgcttaaaa 660 ccaatttgtc cacagtgagt gactgtgtcc accaggtagt ggaacttctg caagagcaga 720 acattgtacc ctatactata atcaaagata tccacgaact ctttgtgccg gaaaacaaac 780 ttgaccacgt ccgagctgag gctgaaactc tcccttcatt atcaattact aagctggatc 840 tccagtgggt ccaggttttg agcgaaggct gggccactcc cctcaaaggt ttcatgcggg 900 agaaggagta cttacaggtt atgcactttg acaccctgct agatgatggc gtgatcaaca 960 tgagcatccc cattgtactg cccgtctctg cagaggataa gacacggctg gaagggtgca 1020 gcaagtttgt cctggcacat ggtggacgga gggtagctat cttacgagac gctgaattct 1080 atgaacacag aaaagaggaa cgctgttccc gtgtttgggg gacaacatgt acaaaacacc 1140 cccatatcaa aatggtgatg gaaagtgggg actggctggt tggtggagac cttcaggtgc 1200 tggagaaaat aagatggaat gatgggctgg accaataccg tctgacacct ctggagctca 1260 aacagaaatg taaagaaatg aatgctgatg cggtgtttgc attccagttg cgcaatcctg 1320 tccacaatgg ccatgccctg ttgatgcagg acacctgccg caggctccta gagaggggct 1380 acaagcaccc ggtcctccta ctacaccctc tgggcggctg gaccaaggat gacgatgtgc 1440 ctctagactg gcggatgaag cagcacgcgg ctgtgctcga ggaaggggtc ctggatccca 1500 agtcaaccat tgttgccatc tttccgtctc ccatgttata tgctggcccc acagaggtcc 1560

RECEIVED

NOV 0 7 2002

TECH CENTER 1600/2900

agtggcactg caggtcccgg atgattgcgg gtgccaattt ctacattgtg gggagggacc 1620 ctgcaggaat gccccatcct gaaaccaaga aggatctgta tgaacccact catgggggca 1680 aggtettgag catggeeect ggeeteacet etgtggaaat cattecatte egagtggetg 1740 cctacaacaa agccaaaaaa gccatqqact tctatgatcc agcaaggcac aatgagtttg 1800 acttcatctc aggaactcga atgaggaagc tcgcccggga aggagagat cccccagatg 1860 getteatgge eeccaaagea tggaaggtee tgacagatta ttacaggtee etggagaaga 1920 actaageett tgggteeaga gtttetttet gaagtgetet ttgattaeet tttetatttt 1980 tatgattaga tgctttgtat taaattgctt ctca <210> 2 <211> 2000 <212> DNA <213> Mus musculus <400>2gtattctcaa catcagatat catgtcttgg aggaagttac ctaaactctg aagaattatc 60 atgtctgcaa atttcaaaat gaaccataaa agagaccagc aaaaatccac caatgtggtc 120 taccaggccc atcatgtgag caggaacaag agaggacaag tggttggaac caggggagga 180 ttccgaggat gtaccgtgtg gctaacaggt ctctctggtg ctgggaaaac aaccataagc 240 tttgctttgg aagagtacct tgtatctcac gccatcccat gttactccct ggatgggac 300 aatgtccgtc atggccttaa taagaacctg ggattctctg ccggggaccg agaagagaat 360 atccgccgga tcgcggaggt ggccaagctc tttgccgacg ccggcctggt ttgcatcacc 420 agetttatet eteettttge aaaggategt gagaatgee gaaaaateea egaateagea 480 qqactcccqt tctttqaqat ctttqtaqat qcqcctttaa atatctqtqa aagccqaqac 540 gtaaaaggac tctacaaacg agcccgagca ggagagatta aagggtttac aggcatcgat 600 tetgaetatg agaaacetga aacteeagag tgtgtgetga agaecaacet gtetteagta 660 agegaetgtg tgeaacaggt ggtggaacet ttgeaggage agaacattgt acceeacace 720 accatcaaag gcatccacga actctttgtg ccagaaaaca aagtcgatca aatccgagct 780 gaggcagaga ctctcccatc actaccaatt accaagctgg atctgcagtg ggtgcagatt 840 ctgagtgaag gctgggccac tcccctcaaa ggctttatgc gggagaagga atacttgcaa 900 actitacact tigacactict actigacigat ggagticatica acatigagitat ticcicattigta 960 ttgcccgttt ctgcggatga caaggcacgg ctcgaagggt gcagcaaatt tgccttgatg 1020 tacgaaggtc ggagggtcgc tctattacag gaccctgaat tctatgagca taggaaagag 1080 gagegttgtt ctcgtgtgtg gggaacagec actgcaaage acceccatat caaaatggtg 1140 atggaaagtg gggactggct tgttggtgga gacctacagg tgctagagag aataaggtgg 1200 gacgatgggc tggaccaata ccgccttacg cctctggaac tcaaacagaa gtgtaaagac 1260 atgaatgctg atgccgtgtt tgcattccag ttgcgcaatc ctgtccacaa tggtcatgcc 1320 ctcctgatgc aggacacccg ccgcaggctc ctggagaggg gttacaagca cccagtcctc 1380 ctgctccacc ctcttggggg ctggaccaag gacgatgacg tacctctgga atggaggatg 1440 aaacagcatg cagctgtact ggaggaaagg gtcctggatc ccaagtcaac tattgttgcc 1500 atettteeat eteetatgtt ataegetggt eecagagg teeagtggea ttgeagatge 1560 cggatgattg caggagccaa tttctacatt gtgggtaggg atcccgcagg aatgccccat 1620 cctgagacaa agaaagacct atatgaaccc acccacgggg gcaaggtctt gagtatggcc 1680 cctggcctta cctctgtgga aataattccg ttccgagtgg ctgcctacaa taaaattaaa 1740 aaggccatgg acttttatga tccagcaagg cacgaggagt ttgacttcat ctcaggaact 1800 cgcatgagga agctcgcccg ggaaggagaa gatcccccag atggcttcat ggccccgaaa 1860

<210> 3 <211> 20 <212> DNA

<213> Homo sapiens

ctgctctgta tccaattgca

gcgtggaaag tgttgacaga ttactacagg tctctggaga agaccaacta ggtgctcctg 1920 gctctggctt cttcctcaag tgctctctga cgatttttt tttctatttt tgtgatttag 1980

		0> 3 acca	agg -	atga	cgat	gt											20
<	21:	0 > 4 1 > 2 2 > D1 3 > He	NA	sapi	ens												
		0> 4 aaag	atg '	gcaa	caat	3 9											20
<	212	0 > 5 1 > 2 2 > Di 3 > Ho	NA	sapi	ens												
		0> 5 gtgc	tgg ,	aaaa	acaa	cg											20
<	212	0> 6 1> 2: 2> Di 3> Ho	NA	sapi	ens	• .											
		0> 6 gaatq	gga (gaaa	taaag	gc to	3										22
<	212	0> 7 l> 6: 2> Pl 3> Ho	RT	sapi	ens												
	let)> 7 Ser	Gly	Ile	Lys	Lys	Gln	Lys	Thr		Asn	Gln	Gln	Lys		Thr	
A	l sn	Val	Va1	Tyr 20	Gln	Ala	His	His	Val 25	10 Ser	Arg	Asn	Lys	Arg 30	15 Gly	Gln	
V	al	Val	Gly 35	Thr	Arg	Gly	Gly	Phe 40		Gly	Cys	Thr	Val	Trp	Leu	Thr	
G	ly	Leu 50	Ser	Gly	Ala	Gly	Lys 55	Thr	Thr	Ile	Ser	Phe 60	Ala	Leu	Glu	Glu	
	'yr 5	Leu	Val	Ser	His	Ala 70	Ile	Pro	Cys	Tyr	Ser 75	Leu	Asp	Gly	Asp	Asn 80	
V	al	.Arg	His	Gly	Leu 85	Asn	Arg	Asn	Leu	Gly 90	Phe	Ser	Pro	Gly	Asp 95	Arg	
G	lu	Glu	Asn	Ile 100	Arg	Arg	Ile	Ala	Glu 105	Val	Ala	Lys	Leu	Phe 110	Ala	Asp	
A	.la	Gly	Leu 115	Val	Cys	Ile	Thr	Ser 120	Phe	Ile	Ser	Pro	Phe 125	Ala	Lys	Asp	
A	.rg	Glu 130	Asn	Ala	Arg	Lys	Ile 135	His	Glu	Ser	Ala	Gly 140		Pro	Phe	Phe	
	lu 45		Phe	Val	Asp	Ala 150		Leu	Asn	Ile	Cys 155		Ser	Arg	Asp	Val 160	
		Gly	Leu	Tyr	Lys 165		Ala	Arg	Ala	Gly 170		Ile	Lys	Gly	Phe 175		

· Gly Ile Asp Ser Asp Tyr Glu Lys Pro Glu Thr Pro Glu Arg Val Leu Lys Thr Asn. Leu Ser Thr Val Ser Asp Cys Val His Gln Val Val Glu Leu Leu Gln Glu Gln Asn Ile Val Pro Tyr Thr Ile Ile Lys Asp Ile His Glu Leu Phe Val Pro Glu Asn Lys Leu Asp His Val Arg Ala Glu Ala Glu Thr Leu Pro Ser Leu Ser Ile Thr Lys Leu Asp Leu Gln Trp Val Gln Val Leu Ser Glu Gly Trp Ala Thr Pro Leu Lys Gly Phe Met Arg Glu Lys Glu Tyr Leu Gln Val Met His Phe Asp Thr Leu Leu Asp Asp Gly Val Ile Asn Met Ser Ile Pro Ile Val Leu Pro Val Ser Ala Glu Asp Lys Thr Arg Leu Glu Gly Cys Ser Lys Phe Val Leu Ala His Gly Gly Arg Arg Val Ala Ile Leu Arg Asp Ala Glu Phe Tyr Glu His Arg Lys Glu Glu Arg Cys Ser Arg Val Trp Gly Thr Thr Cys Thr Lys His Pro His Ile Lys Met Val Met Glu Ser Gly Asp Trp Leu Val Gly Gly Asp Leu Gln Val Leu Glu Lys Ile Arg Trp Asn Asp Gly Leu Asp Gln Tyr Arg Leu Thr Pro Leu Glu Leu Lys Gln Lys Cys Lys Glu Met Asn Ala Asp Ala Val Phe Ala Phe Gln Leu Arg Asn Pro Val His Asn Gly His Ala Leu Leu Met Gln Asp Thr Cys Arg Arg Leu Leu Glu Arg Gly Tyr Lys His Pro Val Leu Leu Leu His Pro Leu Gly Gly Trp Thr Lys Asp Asp Asp Val Pro Leu Asp Trp Arg Met Lys Gln His Ala Ala Val Leu Glu Glu Gly Val Leu Asp Pro Lys Ser Thr Ile Val Ala Ile Phe Pro Ser Pro Met Leu Tyr Ala Gly Pro Thr Glu Val Gln Trp His Cys Arg Ser Arg Met Ile Ala Gly Ala Asn Phe Tyr Ile Val Gly Arg Asp Pro Ala Gly Met Pro His Pro Glu Thr Lys Lys Asp Leu Tyr Glu Pro Thr His Gly Gly Lys Val Leu Ser Met Ala Pro Gly Leu Thr Ser Val Glu Ile Ile Pro Phe Arg Val Ala Ala Tyr Asn Lys Ala Lys Lys Ala Met Asp Phe Tyr Asp Pro Ala Arg His Asn Glu Phe Asp Phe Ile Ser Gly Thr Arg Met Arg Lys Leu Ala Arg Glu Gly Glu Asn Pro Pro Asp Gly Phe Met Ala Pro Lys Ala Trp Lys Val Leu Thr Asp Tyr Tyr Arg Ser Glu Met Asp Lys Asn

<210> 8 <211> 617 <212> PRT <213> Mus musculus

<400> 8 Met Ser Ala Asn Phe Lys Met Asn His Lys Arg Asp Gln Gln Lys Ser Thr Asn Val Val Tyr Gln Ala His His Val Ser Arg Asn Lys Arg Gly Gln Val Val Gly Thr Arg Gly Gly Phe Arg Gly Cys Thr Val Trp Leu Thr Gly Leu Ser Gly Ala Gly Lys Thr Thr Ile Ser Phe Ala Leu Glu Glu Tyr Leu Val Ser His Ala Ile Pro Cys Tyr Ser Leu Asp Gly Asp Asn Val Arg His Gly Leu Asn Lys Asn Leu Gly Phe Ser Ala Gly Asp Arg Glu Glu Asn Ile Arg Arg Ile Ala Glu Val Ala Lys Leu Phe Ala Asp Ala Gly Leu Val Cys Ile Thr Ser Phe Ile Ser Pro Phe Ala Lys Asp Arg Glu Asn Ala Arg Lys Ile His Glu Ser Ala Gly Leu Pro Phe Phe Glu Ile Phe Val Asp Ala Pro Leu Asn Ile Cys Glu Ser Arg Asp Val Lys Gly Leu Tyr Lys Arg Ala Arg Ala Gly Glu Ile Lys Gly Phe Thr Gly Ile Asp Ser Asp Tyr Glu Lys Pro Glu Thr Pro Glu Cys Val Leu Lys Thr Asn Leu Ser Ser Val Ser Asp Cys Val Gln Gln Val Val Glu Leu Leu Gln Glu Gln Asn Ile Val Pro His Thr Thr Ile Lys Gly Ile His Glu Leu Phe Val Pro Glu Asn Lys Val Asp Gln Ile Arg Ala Glu Ala Glu Thr Leu Pro Ser Leu Pro Ile Thr Lys Leu Asp Leu Gln Trp Val Gln Ile Leu Ser Glu Gly Trp Ala Thr Pro Leu Lys Gly Phe Met Arg Glu Lys Glu Tyr Leu Gln Thr Leu His Phe Asp Thr Leu Leu Asp Asp Gly Val Ile Asn Met Ser Ile Pro Ile Val Leu Pro Val Ser Ala Asp Asp Lys Ala Arg Leu Glu Gly Cys Ser Lys Phe Ala Leu Met Tyr Glu Gly Arg Arg Val Ala Leu Leu Gln Asp Pro Glu Phe Tyr Glu His Arg Lys Glu Glu Arg Cys Ser Arg Val Trp Gly Thr Ala Thr Ala Lys His Pro His Ile Lys Met Val Met Glu Ser Gly Asp Trp Leu Val

- 5 -

. Gly Gly Asp Leu Gln Val Leu Glu Arg Ile Arg Trp Asp Asp Gly Leu Asp Gln Tyr.Arg Leu Thr Pro Leu Glu Leu Lys Gln Lys Cys Lys Asp Met Asn Ala Asp Ala Val Phe Ala Phe Gln Leu Arg Asn Pro Val His Asn Gly His Ala Leu Leu Met Gln Asp Thr Arg Arg Arg Leu Leu Glu Arg Gly Tyr Lys His Pro Val Leu Leu His Pro Leu Gly Gly Trp Thr Lys Asp Asp Val Pro Leu Glu Trp Arg Met Lys Gln His Ala Ala Val Leu Glu Glu Arg Val Leu Asp Pro Lys Ser Thr Ile Val Ala Ile Phe Pro Ser Pro Met Leu Tyr Ala Gly Pro Thr Glu Val Gln Trp His Cys Arg Cys Arg Met Ile Ala Gly Ala Asn Phe Tyr Ile Val Gly Arg Asp Pro Ala Gly Met Pro His Pro Glu Thr Lys Lys Asp Leu Tyr Glu Pro Thr His Gly Gly Lys Val Leu Ser Met Ala Pro Gly Leu Thr Ser Val Glu Ile Ile Pro Phe Arg Val Ala Ala Tyr Asn Lys Ile Lys Lys Ala Met Asp Phe Tyr Asp Pro Ala Arg His Glu Glu Phe Asp Phe Ile Ser Gly Thr Arg Met Arg Lys Leu Ala Arg Glu Gly Glu Asp Pro Pro Asp Gly Phe Met Ala Pro Lys Ala Trp Lys Val Leu Thr Asp Tyr Tyr Arg Ser Glu Met Asp Lys Thr Asn

```
<210> 9
<211> 1845
<212> DNA
<213> Homo sapiens
```

<400> 9

 cccgtctctg cagaggataa gacacggctg gaagggtgca gcaagtttgt cctggcacat 960 ggtggacgga gggtagctat cttacgagac gctgaattct atgaacacag aaaagaggaa 1020 cgctgttccc gtgtttgggg gacaacatgt acaaaacacc cccatatcaa aatqgtgatg 1080 gaaagtgggg actggctggt tggtggagac cttcaggtgc tggagaaaat aagatggaat 1140 gatgggetgg accaataceg tetgacacet etggagetea aacagaaatg taaagaaatg 1200 aatgctgatg cggtgtttgc attccagttg cgcaatcctg tccacaatgg ccatgccctg 1260 ttgatgcagg acacctgccg caggctccta gagaggggct acaagcaccc ggtcctccta 1320 ctacaccete tgggeggetg gaccaaggat gacgatgtge etetagactg geggatgaag 1380 cagcacgegg ctgtgctcga ggaaggggtc ctggatccca agtcaaccat tgttgccatc 1440 tttccgtctc ccatgttata tgctggcccc acagaggtcc agtggcactg caggtcccgg 1500 atgattgcgg gtgccaattt ctacattgtg gggagggacc ctgcaggaat gccccatcct 1560 gaaaccaaga aggatetgta tgaacccaet catgggggca aggtettgag catggeeeet 1620 ggcctcacct ctgtggaaat cattccattc cgagtggctg cctacaacaa agccaaaaaa 1680 gccatggact tctatgatcc agcaaggcac aatgagtttg acttcatctc aggaactcga 1740 atgaggaagc tcgcccggga aggagagaat cccccagatg gcttcatggc ccccaaagca 1800 tggaaggtcc tgacagatta ttacaggtcc ctggagaaga actaa <210> 10 <211> 1851 <212> DNA <213> Mus musculus <400> 10

atgtctgcaa atttcaaaat gaaccataaa agagaccagc aaaaatccac caatgtggtc 60 taccaggccc atcatgtgag caggaacaag agaggacaag tqqttggaac caggqgagqa 120 ttccgaggat gtaccgtgtg gctaacaggt ctctctggtg ctgggaaaac aaccataagc 180 tttgctttgg aagagtacct tgtatctcac gccatcccat gttactccct ggatggggac 240 aatgtccgtc atggccttaa taagaacctg ggattctctg ccggggaccg agaagagaat 300 atccgccgga tcgcggaggt ggccaagctc tttgccgacg ccggcctggt ttgcatcacc 360 agetttatet eteettttge aaaggategt gagaatgee gaaaaateea egaateagea 420 ggactcccgt tetttgagat etttgtagat gegeetttaa atatetgtga aageegagae 480 gtaaaaggac tetacaaacg ageeegagea ggagagatta aagggtttac aggeategat 540 tetgaetatg agaaacetga aacteeagag tgtgtgetga agaecaaett gtetteagta 600 agegaetgtg tgeaacaggt ggtggaactt tigeaggage agaacattgt accecacace 660 accatcaaag gcatccacga actctttgtg ccagaaaaca aagtcgatca aatccgagct 720 gaggcagaga ctctcccatc_actaccaatt_accaagctgg atctgcagtg ggtgcagatt 780 ctgagtgaag gctgggccac tcccctcaaa ggctttatgc gggagaagga atacttgcaa 840 actctacact tcgacactct actggacgat ggagtcatca acatgagtat tcccattgta 900 ttgcccgttt ctgcggatga caaggcacgg ctcgaagggt gcagcaaatt tgccttgatg 960 tacgaaggtc ggagggtcgc tctattacag gaccctgaat tctatgagca taggaaagag 1020 gagegttgtt etegtgtgtg gggaacagee aetgeaaage aeceecatat caaaatggtg 1080 atggaaagtg gggactggct tgttggtgga gacctacagg tgctagagag aataaggtgg 1140 gacgatgggc tggaccaata ccgccttacg cctctggaac tcaaacagaa gtgtaaagac 1200 atgaatgetg atgeegtgtt tgeatteeag ttgegeaate etgteeaeaa tggteatgee 1260 ctcctgatgc aggacacccg ccgcaggctc ctggagaggg gttacaagca cccagtcctc 1320 ctgctccacc ctcttggggg ctggaccaag gacgatgacg tacctctgga atggaggatg 1380 aaacagcatg cagctgtact ggaggaaagg gtcctggatc ccaagtcaac tattgttgcc 1440 atctttccat ctcctatgtt atacgctggt cccacagagg tccagtggca ttgcagatgc 1500 cggatgattg caggagccaa tttctacatt gtgggtaggg atcccgcagg aatgccccat 1560 cctgagacaa agaaagacct atatgaaccc acccacgggg gcaaggtctt gagtatggcc 1620 cctggcctta cctctgtgga aataattccg ttccgagtgg ctgcctacaa taaaattaaa 1680 aaggccatgg acttttatga tccagcaagg cacgaggagt ttgacttcat ctcaggaact 1740 cgcatgagga agctcgcccg ggaaggagaa gatcccccag atggcttcat ggccccgaaa 1800

1851

gcgtggaaag tgttgacaga ttactacagg tctctggaga agaccaacta g

<210> 11 <211> 21 <212> DNA <213> Homo	sapiens		
<400> 11 gccagccagc	atgtcgggga	t	21
<210> 12 <211> 24 <212> DNA <213> Homo	sapiens		
<400> 12 acctgaaact	cctgagcgtg	tgct	24
<210> 13 <211> 21 <212> DNA <213> Homo	sapiens		
<400> 13 gatgtgcctc	tagactggcg	g	21
<210> 14 <211> 24 <212> DNA <213> Homo	sapiens		
<400> 14 gagcacttca	gaaagaaact	ctgg	24
<210> 15 <211> 21 <212> DNA <213> Homo	sapiens		
<400> 15 catccgccag	tctagaggca	c .	21
<210> 16 <211> 21 <212> DNA <213> Homo	sapiens		
<400> 16 aggtgtcaga	cggtattggt	c	21
<210> 17 <211> 23 <212> DNA <213> Homo	sapiens		
<400> 17 gtcactcact	gtggacaaat	tgg	23

<210> 18 <211> 21 <212> DNA <213> Homo sapiens		
<400> 18 cacctcagca atccggcgga	t	21
<210> 19 <211> 20 <212> DNA <213> Mus musculus		
<400> 19 tctggcacaa agagttcgtg		20
<210> 20 <211> 22 <212> DNA <213> Mus musculus		
<400> 20 gccagtttgt aaccgagtat	tc	22
<210> 21 <211> 22 <212> DNA <213> Mus musculus		
<400> 21 gcaattggat acagagcagc	ta	22
<210> 22 <211> 22 <212> DNA <213> Mus musculus		
<400> 22 gacaatgtcc gtcatggcct	ta	22
<210> 23 <211> 21 <212> DNA <213> Mus musculus		
<400> 23 attcccattg tattgcccgt	t	21
<210> 24 <211> 21 <212> DNA <213> Mus musculus		
<400> 24		

aacgggcaat acaatgggaa t	21
<210> 25 <211> 22 <212> DNA <313> Myg myggylyg	
<213> Mus musculus <400> 25 gataaagetg gtgatgeaaa ee	22
<210> 26 <211> 20 <212> DNA <213> Mus musculus	
<400> 26 catgggatgg cgtgagatac	20
<210> 27 <211> 23 <212> DNA <213> Mus musculus	
<400> 27 cataagcttt gctttggaag agt	23
<210> 28 <211> 21 <212> DNA <213> Homo sapiens	
<400> 28 gcatgtccag acagacacca c	21
<210> 29 <211> 333 <212> DNA <213> Homo sapiens	
<220> <223> D19Mit13 locus and flanking sequences	
<221> misc_feature <222> (1)(333) <223> n = A,T,C or G; at nucleotide positions 23 and 305	
<pre><400> 29 ctgactatga gaaacctgaa acnccagagt gtgtgctgaa gaccaacctg tcttcagtaa gcgactgtgt gcaacaggtg gtggaacttt tgcaggagca ggtaggaggg tggttcttgc cagtgtgttc agtgtgtgtg tgtgtgtgtg tgtgtgtgtg tgcgtgtgca tgtgtgtgtg catgtgtgtg tgcgtgtgca tgtgtgtgtg ttgaaagata atctgagttt ctttattccc tggccaatct cagtaactat tgccaatttc gtttcccaca gaacattgta cccacacca ccatnaaagg catccacgaa ctctttgtgc cag</pre>	120 180 240

<210> 30

```
<211> 18
 <212> DNA
 <213> Homo sapiens
 <220>
 <223> Nucleotide positions +1414 through +1431 of PAPSS2
       coding sequence
 <400> 30
 gatcccaagt caaccatt
                                                                     18
 <210> 31
 <211> 6
 <212> PRT
 <213> Homo sapiens
 <220>
 <223> Partial PAPSS2 peptide sequence; amino acid
       residues 472 through 477
 <400> 31
 Asp Pro Lys Ser Thr Ile
 <210> 32
 <211> 18
 <212> DNA
 <213> Homo sapiens
 <220>
 <221> mutation
 <222> (0) ...(0)
 <223> Nucleotide positions +1414 through +1431 of PAPSS2
       coding sequence with mutation c to a at nucleotide
       position +1424
 <400> 32
 gatcccaagt aaaccatt
                                                                     18
 <210> 33
 <211> 3
 <212> PRT
 <213> Homo sapiens
 <223> Partial truncated PAPSS2 peptide sequence; amino
       acid residues 472-474 plus stop at position 475
```

- 11 -

<400> 33 Asp Pro Lys

1